**PANELISTS AND CONFIRMED SPEAKERS**

# Advisory Panel on Rare Disease

## Spring Meeting: May 18, 2022

**GotoWebinar**

### Rare Disease Advisory Panel Members

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**Mathew Edick, PhD**
Chair, Advisory Panel on Rare Disease
Stakeholder Affiliation: Patients, Caregivers, and Patient Advocates

Director of the Center for Strategic Health Partnerships, Michigan Public Health Institute

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*Term Ends August 2023*

Mathew J. Edick is the Director of the Center for Strategic Health Partnerships at Michigan Public Health Institute. He is an organizational PCORI Ambassador and has been a principal investigator, co-investigator, project manager, and programmatic/technical assistance lead on four PCORI-funded projects. Edick works with local, regional, and national-scale projects that engage diverse stakeholders to identify and implement innovative solutions for improving health outcomes. He received his doctorate in pharmacogenomics from the University of Tennessee.

**Doug Lindsay, BS**
Co-Chair, Advisory Panel on Rare Disease
Stakeholder Affiliation: Patients, Caregivers and Patient Advocates

Personal Medical Consultant and Founder, Doug Says LLC.

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*Term Ends August 2022*

Doug Lindsay became ill at age 21 and spent the next 11 years home- and bedbound until he figured out what was wrong, developed novel treatments to keep his rare autonomic-adrenal condition at bay, and eventually developed the innovative surgery used to treat him.

During Lindsay's 14-year medical odyssey, he worked with 35 senior faculty at 28 institutions, developed new uses for five existing prescription drugs, won a national first court case protecting patients' rights, and developed the concept for the two successful, innovative adrenal surgeries used to treat his case.

Now, Lindsay is a nationally recognized speaker on the role of hope and character in innovation and in life. He is a Community Advisory Board member for Washington University's Institute of Clinical and Translational Sciences and Institute for Public Health. He also provides an in-depth personal medical consultant service to a small number of rare disease and complex condition patients who have found themselves trapped in the medical system.

**Heather Adams, PhD**
Member, Advisory Panel on Rare Disease
Stakeholder affiliation: Researchers

Associate Professor, University of Rochester Medical Center

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*Term Ends August 2024*

Heather Adams is an Associate Professor in the Department of Neurology at the University of Rochester Medical Center. She is a member of the University of Rochester Batten Center (URBC) and has previously served on the Medical Advisory Board (MAB) to the Batten Disease Support & Research Association, the major US advocacy group for individuals impacted by Batten disease.

Dr. Adams' research includes a focus on clinical endpoints, quality of life, and the everyday impacts experienced by individuals with rare, inherited metabolic and neurodegenerative disorders, their parents/caregivers, and siblings. Her research interests also include cognitive function in pediatric hypertension, HIV-associated neurocognitive disorders, and improved screening and diagnosis of Tourette syndrome and other tic disorders. Her clinical activities include neuropsychological assessment and evidence-based psychosocial interventions for children.

Dr. Adams' rare disease research has always been informed by the voice of patients and their parents and families. Since joining the URBC in 2003, she has actively engaged with the BDSRA as a researcher to develop studies, implement recruitment/engagement activities, and share research results with the BDSRA and its members. As a clinical child psychologist and pediatric neuropsychologist, she is committed to bringing forward each child's voice, and hopes to apply her perspective and expertise in particular to the consideration of pediatric patient-centered outcomes research as a member of the PCORI Advisory Panel on Rare Disease.

**Sarah Bacon, MS**
Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Patients, Caregivers and Patient Advocates

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*Term Ends August 2023*

Sarah Bacon is a rare disease patient, advocate, and writer. Shortly after her 2013 diagnosis with lymphangioleiomyomatosis (LAM), and after the sequester cuts to the National Institutes of Health, she wrote an op-ed in The Atlantic, "Medical Research Cuts Have Immediate Health Effects." Since then, she has applied her 15 years of professional communications, advocacy, and issue-related campaign design experience to advancing rare disease research and policy.

Bacon suggested and raised funds for a novel study on the link between LAM and melanoma, which share several biomarkers, including proteins. Melanoma took her father's life and threatened hers. Fortunately, hers was caught early. The study bore meaningful results on the high incidence of melanoma family histories in LAM patients. When the LAM Lab faced losing a commitment from Novartis of a $200,000 in-kind donation for a pilot trial, Bacon advocated with Novartis executives and restored the donation.

Bacon secured bipartisan sponsors of The OPEN Act in the House of Representatives with Rare Disease Legislative Advocates, spoke on patient-driven medicine for Global Genes and The Milken Institute, and served on Global Genes' grant application review committee. She has written on rare diseases for The Atlantic, Fast Company, The Washington Post, and New York Magazine, and is currently working on her first book, Living with Zebras.

**Danielle Boyce, MPH**
Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Patients, Caregivers, and Patient Advocates

Senior Research Data Analyst, COPD Foundation

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*Term Ends August 2023*

Danielle Boyce is an award-winning rare epilepsy advocate, researcher, public speaker, and writer. Her work has appeared in dozens of scientific journals and her children's book, Charlie's Teacher, is used in children's hospitals throughout the country. She has served on several patient and caregiver advisory panels for academic institutions, pharmaceutical companies, nonprofits, the Food and Drug Administration, the American Academy of Pediatrics, and PCORI. Boyce currently works for the COPD Foundation and advises rare disease organizations on data science-related matters. She previously worked for Johns Hopkins Schools of Medicine and Nursing. She holds a master's in public health with a concentration in epidemiology and is currently working toward a doctorate in public administration.

After representing the caregiver voice on a PCORI-funded study, Boyce realized that serving on a PCORI advisory panel is the perfect marriage of her research and advocacy skills and is excited to serve patients through her continued involvement with PCORI. She lives outside of Philadelphia with her husband, Jim, and their four children.

**Natario Couser, MD**
Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Clinicians

Assistant Professor of Opthalmology, Pediatrics, and Human & Molecular Genetics, Virginia Commonwealth University

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*Term Ends August 2024*

Natario Couser received his undergraduate degree in biochemistry from the University of Virginia and completed his medical education at the Virginia Commonwealth University (VCU) School of Medicine. Couser completed his ophthalmology residency at Howard University, serving as co-chief resident in his final year. Couser received fellowship training in pediatric ophthalmology and adult strabismus at Emory University, completed a Master of Science degree in biotechnology from Johns Hopkins University and training in clinical genetics at the University of North Carolina at Chapel Hill.

Couser is one of only a few individuals currently board-certified by both the American Board of Ophthalmology and the American Board of Medical Genetics and Genomics. Couser has served as the principal investigator for clinical trials, presented invited lectures, won awards for research, and has published numerous articles and book chapters on rare genetic diseases, and served as the editor of the book Ophthalmic Genetic Diseases.

Couser is currently an Assistant Professor with the Departments of Ophthalmology, Pediatrics and Human & Molecular Genetics at VCU in Richmond, Virginia, where he initiated the first dedicated ophthalmic genetics specialty service in the state. PCORI's devotion to clinical effectiveness, engagement, and research infrastructure involving rare diseases with a patient-centered approach is what motivated Couser to strive to support their ongoing work in this area.

**Giovanna Devercelli, PhD**
Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Industry

Head, Rare Diseases, Global Evidence and Outcomes, Takeda Pharmaceuticals

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*Term Ends August 2024*

Giovanna Devercelli serves as Executive Director, Head, Global Evidence and Outcomes Research at Takeda where she leads the Rare Diseases and Plasma Derived Therapies Therapeutic Areas, responsible for creating and executing integrated real-world data/evidence (RWD/E) and clinical outcome assessments (COAs) strategic plans and tactics through evidence generation including patient-centric outcomes (e.g., patient-reported endpoints).

Devercelli possesses over 20 years' experience spanning clinical research and development, market access, real-world evidence generation (observational study design and execution), clinical outcomes assessments, and health economics. Devercelli has supported numerous regulatory and health technology submissions for products in various therapeutic areas.

Devercelli completed her PhD at the University of Louisiana at Monroe. Born in Lima, Peru, Devercelli loves to play tennis, and has a 16-year-old daughter.

**Deanna Fournier, BS**
Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Patients, Caregivers, and Patient Advocates

Executive Director, Histiocytosis Association

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*Term Ends August 2024*

Deanna Fournier is the Executive Director of the Histiocytosis Association. Fournier's passion and dedication stems from her own diagnosis with Langerhans cell histiocytosis at the age of 6, and her lifetime involvement with the organization.

Fournier brings her experience as a former Director of Business Development, Manager of Global Learning & Development, Wilderness EMT, and Internationally Certified Health Coach to the association in addition to her knowledge and expertise in the medical and rare disease space. Fournier served on the association's Board of Trustees for two years prior to taking on the role of Executive Director.

Fournier has the unique position of being a patient, an organization leader, and an advocate for wellness and health. Fournier is excited and motivated to help PCORI by leveraging her unique perspective to help ensure patients and families have the resources they need to make informed decisions for their health care and to support research that seeks to improve patient outcomes.

**Salman Hussain, MPH**
Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Industry

Consultant, Charles River Associates

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*Term Ends August 2023*

Salman Hussain is a life sciences consultant at Charles River Associates (CRA), and a member of the Board of Directors for the National Alopecia Areata Foundation (NAAF). He has more than seven years of experience in healthcare research and has advised leading pharmaceutical and biotechnology companies across many therapeutic areas (with a specific focus in rare disease), both globally and in the United States.

Prior to joining CRA, Hussain conducted research at Massachusetts General Hospital and The Dartmouth Institute for Health Policy & Clinical Practice, specializing in the rare disease alopecia areata universalis and the interface between dermatology and mental health. While working with NAAF on a PCORI-funded project, he developed a patient-centered outcomes research and comparative clinical effectiveness research training program for patients. He was so inspired after this project that he became a PCORI Ambassador and has continued to advise his pharmaceutical clients and academic researchers on how they can meaningfully engage with patients to inform their research.

In addition to his work in alopecia areata, Hussain has advised and conducted substantial research for pharmaceutical companies to better understand the patient experience in many rare diseases, such as Duchene muscular dystrophy, spinal muscular atrophy types 3 and 4, acute lymphoblastic leukemia, maple syrup urine disease, nemaline myopathy, GM3-synthase deficiency, aromatic 1-amino acid decarboxylase deficiency, and acute hepatic porphyria.

**Sonia Jain, PhD**
Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Researchers

Professor of Biostatistics, University of California, San Diego

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*Term Ends August 2024*

Sonia Jain is a Professor of Biostatistics at the Herbert Wertheim School of Public Health and Human Longevity Science at the University of California, San Diego.

Jain received her PhD in Statistics from the University of Toronto in Toronto, Canada. Her methodological statistical interests include Bayesian biostatistics, with special emphasis on nonparametric Bayesian models. Her research interests include adaptive clinical trial designs and precision medicine trial designs.

Jain is the lead biostatistician for many collaborative projects in several disciplines, including HIV/AIDS, infectious diseases, pediatric cardiology, posttraumatic stress disorder, and traumatic brain injury.

**Mileva Repasky, MS**
Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Patients, Caregivers, and Patient Advocates

Chief Patient Officer, Castleman Disease Collaborative Network

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*Term Ends August 2024*

Mileva Repasky is the Chief Patient and Development Officer for the Castleman Disease Collaborative Network (CDCN) and (most importantly) mother to one of the youngest Castleman disease patients, Katie. Repasky is passionate about connecting patients, family and friends; fundraising; patient advocacy; and raising awareness.

Repasky will continue to advance the PCORI and CDCN missions by collaborating with the patient, family and friends community; physicians; researchers; various stakeholders; and partners. Repasky is particularly interested in finding new and innovative ways to raise awareness for rare diseases.

Repasky was the Project Engagement Lead for CDCN's partnership with the Chan Zuckerberg Initiative in 2019, 2020, and ongoing work on drug repurposing.

Repasky graduated from Duquesne University with a BS in Psychology and completed her Master of Science in Psychology with a concentration in Behavioral Health in 2016.

**Bridget Reynolds**
Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Patients, Caregivers, and Patient Advocates

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*Terms Ends August 2024*

Bridget Reynolds is a member of Improving Sickle Cell Care in Adolescents and Adults in Chicago (ISAAC) patient advisory council and CRiSP. Reynolds has served as a team leader in the University of Illinois iPartner study, chaired the Member Advisory Council for an HMO.

Reynolds has worked as a Certified Medical Technologist in public and private health care. She served as a Managing Supervisor of group homes for the developmentally disabled, a subcontract with the Illinois Department of Public Health, on minority health initiatives.

Reynolds writes poetry, is an artist, and has been a critic and feature editor for a local newspaper.

Reynolds dreams of making modalities, including artificial intelligence helper bots and humanoid robots, accessible to persons with sickle cell disease. Reynolds is confident that the SCD community would be greatly accommodated in everyday life or while in crisis by helper robots. Reynolds feels robots would be especially helpful to those who live alone or care for children without a partner. They have the potential to save lives as the search for a cure continues.

**Nancy Rose, MD**

Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Clinicians

Adjunct Professor of Obstetrics and Gynecology,
University of Utah

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*Term Ends August 2023*

Currently a Medical Consultant at the National Coordinating Center for the Regional Genetics Networks, Nancy Rose was previously the Director of Reproductive Genetics at Intermountain Healthcare in Salt Lake City Utah for 16 years, with the goal to decrease healthcare disparities and disseminate information prior to and during pregnancy as well as in the newborn period throughout the state. She has served as the President of the American Board of Medical Genetics and Genomics and Chair of the Committee on Genetics for the American College of Obstetricians and Gynecologists, and continues to serve on committees for the American College of Medical Genetics and Genomics, the American College of Obstetricians and Gynecologists, the American Board of Medical Genetics and Genomics, and the American Board of Medical Specialties. She remains an adjunct Professor of Obstetrics and Gynecology at the University of Utah.

**Jasvinder Singh, MD, MPH**
Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Researchers

Professor of Medicine, University of Alabama at Birmingham

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*Terms Ends August 2024*

Jasvinder Singh is Professor of Medicine and Epidemiology and holds the Musculoskeletal Outcomes Research Endowed Professor of Medicine. Singh has served as Director of the Rheumatology Research at the Birmingham VA Medical Center since 2009. Singh has also been the Director of the University of Alabama at Birmingham's Cochrane Musculoskeletal Group Satellite Center since 2013; and the Director of the University of Alabama Health Services Foundation gout clinic since 2014.

Singh currently serves on the Food and Drug Administration's Arthritis Advisory Committee, and on the executive committee of Outcomes Measures in Rheumatology Trials. Singh is the current chair of the Department of Veterans Affairs Rheumatology Field Advisory Committee and the Chair of the Health Services Research Grant Review Committee of the Rheumatology Research Foundation.

Singh's research has been continuously funded by federal agencies (National Institutes of Health, Department of Veterans Affairs, Patient-Centered Outcomes Research Institute [PCORI]) and is focused largely on outcomes of arthritis and musculoskeletal diseases. Singh has authored more than 550 manuscripts, 20 book chapters, and 34 editorials.

Singh serves as investigator on multiple research grants focused on patient-reported outcomes, health disparities, and comparative effectiveness research in gout, osteoarthritis, lupus, rheumatoid arthritis, and total joint replacement. Singh has collaborated with colleagues across several disciplines such as orthopedic surgery, rehabilitation, genetics, and oncology.

**Saira Sultan, JD**

Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Policy Makers

CEO, Connect4Strategies

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*Term Ends August 2022*

Saira Sultan, JD, has been at the nexus of patient groups, the medical community, drug/device manufacturers, and governmental organizations, for more than two decades. Her passion for rare and ultrarare disease issues culminated in her founding of the Haystack Project in 2016. Through Haystack, Sultan has given voice to the experiences and perspectives of patients with rare and ultrarare conditions to influence research and policy making. She also continues to work with patient groups suffering from more common conditions such as cancer, sickle cell disease, pain, and addiction as the President and CEO of Connect 4 Strategies, a boutique reimbursement, value, and access consulting firm.

Her history with the cancer community stretches back nearly 20 years to when she directed government affairs for the Association of Community Cancer Centers. During her career, she also held leadership positions with Pfizer, Sanofi, and Medtronic. Sultan holds a juris doctor from the University of Virginia in Charlottesville, Virginia.

**Laura Tosi, MD**

Member, Advisory Panel on Rare Disease
Stakeholder Affiliation: Clinicians

Founder and Director of the Bone Health Program, Children's National Hospital

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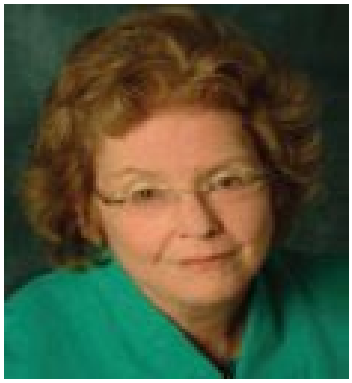
*Term Ends August 2023*

Laura L. Tosi serves on the Board of Directors of the Osteogenesis Imperfecta Foundation (OIF) and the US Bone & Joint Initiative. She serves as site principal investigator for the Brittle Bone Disease Consortium's Osteogenesis Imperfecta Longitudinal Study and has particularly supported the study's patient-reported outcomes research. Tosi served as the principal investigator for the OIF's first patient centered outcomes research project and is currently serving as a co-lead for an OIF PCORI-funded project on improving patient-centered outcomes. She is chair of the Rare Bone Disease Alliance Steering Committee and faculty chair of the Rare Bone Disease TeleECHO program.

Tosi is the Founder and Director of the Bone Health Program at Children's National Hospital and Associate Professor of orthopaedics and pediatrics at George Washington University in Washington, D.C. Her clinical practice focuses on the orthopedic care of children and young adults with physical disabilities and/or issues related to bone health/rare skeletal disease. She is a graduate of Harvard Medical School and received her orthopaedic training at the Columbia Presbyterian Hospital in New York and the Hospital for Sick Children in Toronto.



**Naomi Aronson, PhD**

Ex-Officio Member from PCORI's Methodology Committee

Executive Director, Clinical Evaluation, Innovation, and Policy, Blue Cross Blue Shield Association (BCBSA)

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Dr. Naomi Aronson leads BCBSA clinical effectiveness and policy engagement with government, regulatory agencies, and policy consortia. Her areas of leadership include comparative effectiveness, patient centered research, safety surveillance, regulatory science, and methodological standards. Previously, Dr. Aronson led the development of the BCBSA Technology Evaluation Center (TEC), now Evidence Street™ (ES), as a nationally recognized technology assessment program and an Evidence- based Practice Center (EPC) of the Agency for Healthcare Research and Quality (AHRQ).

She is a member of the Methodology Committee of the Patient-Centered Outcomes Research Institute (PCORI), the Health Technology Assessment International Health Policy Forum, the National Academy of Medicine Genomics Roundtable, the New Drug Development Paradigms (NEWDIGS) initiative of the MIT Center for Biomedical Innovation, and the Medical Device Innovation Consortium (MDIC) National Evaluation System for Health Technology (NEST) Governing Committee.

Prior to joining BCBSA, Dr. Aronson was a member of the Northwestern University faculty, specializing in the sociology of science and medicine. She also was a post-doctoral fellow in the Science, Technology and Society Program at the Massachusetts Institute of Technology.

# PCORI RDAP Staff Team

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# RDAP Spring 2022 Meeting Presenters

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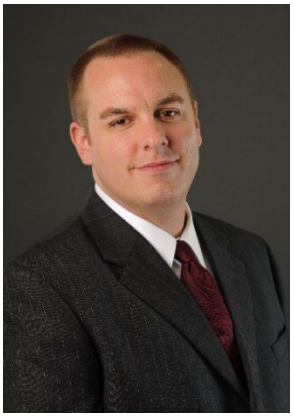

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